



3p deletion syndrome

3p deletion syndrome is a condition that results from a chromosomal change in which a small piece of chromosome 3 is deleted in each cell. The deletion occurs at the end of the short (p) arm of the chromosome. This chromosomal change often leads to intellectual disability, developmental delay, and abnormal physical features.

Individuals with 3p deletion syndrome typically have severe to profound intellectual disability. Most have delayed development of language skills as well as motor skills such as crawling and walking. While affected individuals learn to walk in childhood, their language ability usually remains limited. Some individuals with 3p deletion syndrome have obsessive-compulsive disorder (OCD) or features of autism spectrum disorders, which are conditions characterized by impaired communication and social interaction.

The physical signs and symptoms of 3p deletion syndrome vary greatly. Many affected individuals have slow growth, an abnormally small head (microcephaly), a small jaw (micrognathia), droopy eyelids (ptosis), malformed ears or nose, and widely spaced eyes (hypertelorism). Other frequent features include skin folds covering the inner corner of the eyes (epicanthal folds), extra fingers or toes (polydactyly), and an opening in the roof of the mouth (cleft palate). Additionally, individuals with 3p deletion syndrome may have seizures, weak muscle tone (hypotonia), intestinal abnormalities, or congenital heart defects.

Frequency

3p deletion syndrome is likely a rare disorder; at least 30 cases have been described in the scientific literature.

Genetic Changes

3p deletion syndrome is caused by deletion of the end of the small (p) arm of chromosome 3. The size of the deletion varies among affected individuals, ranging from approximately 150,000 DNA building blocks (150 kilobases or 150 kb) to 11 million DNA building blocks (11 megabases or 11 Mb). The deletion can include between 4 and 71 known genes. In some individuals, the deletion involves material near the end of the chromosome but does not include the tip (the telomere).

The signs and symptoms related to 3p deletion syndrome result from the loss of genes in the 3p region. It is difficult to determine which genes may be responsible for which specific features of 3p deletion syndrome because of the variability in both the size of the deletion and in the signs and symptoms of the condition among affected individuals. Multiple genes at the end of chromosome 3 appear to play a role in neurological development, but because not all people with 3p deletion syndrome are missing the

same genes, it is difficult to pinpoint which ones influence the cognitive symptoms. It is likely that the loss of multiple genes contribute to the different physical abnormalities.

Inheritance Pattern

Most cases of 3p deletion syndrome are not inherited. The deletion occurs in one chromosome, most often as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development. In these cases, affected people have no history of the disorder in their family.

In rare cases, 3p deletion syndrome is inherited, usually from a mildly affected parent. The deletion can also be inherited from an unaffected parent who carries a chromosomal rearrangement between chromosome 3 and another chromosome. This rearrangement is called a balanced translocation. No genetic material is gained or lost in a balanced translocation, so these chromosomal changes usually do not cause any health problems. However, translocations can become unbalanced as they are passed to the next generation. Children who inherit an unbalanced translocation have a chromosomal rearrangement with extra or missing genetic material. Individuals with 3p deletion syndrome associated with an unbalanced translocation are missing genetic material from the short arm of chromosome 3, which results in the signs and symptoms of this disorder.

Other Names for This Condition

- 3p partial monosomy syndrome
- 3p- syndrome
- chromosome 3, deletion 3p
- chromosome 3, monosomy 3p
- chromosome 3p deletion syndrome
- del(3p) syndrome
- deletion 3p
- monosomy 3p
- partial monosomy 3p

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Chromosome 3, monosomy 3p
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0795806/>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

Genetic and Rare Diseases Information Center

- Chromosome 3p deletion
<https://rarediseases.info.nih.gov/diseases/37/chromosome-3p-deletion>
- Chromosome 3p- syndrome
<https://rarediseases.info.nih.gov/diseases/3750/chromosome-3p--syndrome>

Educational Resources

- Centers for Disease Control and Prevention: Facts About Intellectual Disability
<https://www.cdc.gov/ncbddd/developmentaldisabilities/facts.html>
- Centers for Disease Control and Prevention: Facts About Microcephaly
<https://www.cdc.gov/ncbddd/birthdefects/microcephaly.html>
- Disease InfoSearch: 3p deletion syndrome
<http://www.diseaseinfosearch.org/3p+deletion+syndrome/9531>
- Kaiser Permanente: Cleft Lip/Cleft Palate
<https://mydoctor.kaiserpermanente.org/ncal/specialty/genetics/resources/conditions/cleft.jsp>
- Kennedy Krieger Institute: Intellectual Disability
<https://www.kennedykrieger.org/patient-care/diagnoses-disorders/intellectual-disability>
- MalaCards: chromosome 3p- syndrome
http://www.malacards.org/card/chromosome_3p_syndrome
- Merck Manual Consumer Version: Intellectual Disability
<http://www.merckmanuals.com/home/children-s-health-issues/learning-and-developmental-disorders/intellectual-disability>

- Orphanet: Distal monosomy 3p
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1620
- Unique: 3p25 Deletions
<http://www.rarechromo.org/information/Chromosome%20%203/3p25%20deletions%20FTNW.pdf>
- Unique: 3p26 Deletions
<http://www.rarechromo.org/information/Chromosome%20%203/3p26%20deletions%20FTNW.pdf>

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/chromosome-3-monosomy-3p/>
- Unique: The Rare Chromosome Disorder Support Group
<http://www.rarechromo.org/html/home.asp>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%283p+deletion+syndrome%5BTIAB%5D%29+OR+%28monosomy+3p%5BTIAB%5D%29+OR+%28deletion+3p%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- CHROMOSOME 3pter-p25 DELETION SYNDROME
<http://omim.org/entry/613792>

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